EP300 gene

E1A binding protein p300

Normal Function

The *EP300* gene provides instructions for making a protein called p300. This protein regulates the activity of many genes in tissues throughout the body. It plays an essential role in controlling cell growth and division and prompting cells to mature and assume specialized functions (differentiate). The p300 protein appears to be critical for normal development before and after birth.

The p300 protein carries out its function by activating transcription, the process of making a blueprint of a gene for protein production. Specifically, p300 connects transcription factors, which are proteins that start the transcription process, with the complex of proteins that carries out transcription. On the basis of this function, p300 is called a transcriptional coactivator.

Health Conditions Related to Genetic Changes

prostate cancer

Rubinstein-Taybi syndrome

Several mutations in the *EP300* gene have been identified in people with Rubinstein-Taybi syndrome. These genetic changes are responsible for a small percentage of cases of this condition. Some mutations lead to the production of an abnormally small, nonfunctional version of the p300 protein, while other mutations prevent one copy of the gene from making any protein at all. These genetic changes all result in the loss of one functional copy of the *EP300* gene in each cell, which reduces the amount of p300 protein by half. Although researchers are uncertain how a reduction in the amount of this protein leads to the specific features of Rubinstein-Taybi syndrome, it is clear that the loss of one copy of the *EP300* gene disrupts normal development before and after birth.

cancers

Rarely, chromosomal rearrangements (translocations) involving chromosome 22 have been associated with certain types of cancer. These genetic changes are somatic, which means they are acquired during a person's lifetime and are present only in certain cells. In cancer cells, translocations can disrupt the region of chromosome 22 that contains the *EP300* gene. For example, researchers have found a translocation between chromosome 8 and chromosome 22 in several people

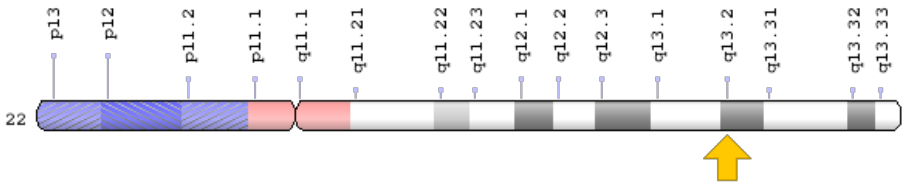
with a cancer of blood-forming cells called acute myeloid leukemia (AML). Another translocation, involving chromosomes 11 and 22, has been found in a small number of people who have undergone cancer treatment. This chromosomal change is associated with the development of AML following chemotherapy for other forms of cancer.

Somatic mutations in the *EP300* gene have been identified in several other types of cancer. These mutations prevent the gene from producing any functional protein. Cells without the p300 protein cannot effectively restrain growth and division, allowing cancerous tumors to develop and grow. Somatic mutations in the *EP300* gene have been found in a small number of solid tumors, including cancers of the colon and rectum, stomach, breast, and pancreas. Studies suggest that *EP300* mutations may also play a role in the development of some prostate cancers. These genetic changes could help predict whether prostate tumors will increase in size or spread to other parts of the body.

Chromosomal Location

Cytogenetic Location: 22q13.2, which is the long (q) arm of chromosome 22 at position 13.2

Molecular Location: base pairs 41,092,610 to 41,180,077 on chromosome 22 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- E1A-associated protein p300
- E1A-binding protein, 300kD
- EP300_HUMAN
- p300
- p300 E1A-Associated Coactivator

Additional Information & Resources

Educational Resources

- Cancer Medicine (sixth edition, 2003): Chromosomal Abnormalities Involving Transcriptional Coactivators
  https://www.ncbi.nlm.nih.gov/books/NBK12465/#A44443
- Molecular Biology of the Cell (fourth edition, 2002): Eucaryotic Gene Regulatory Proteins Often Assemble into Complexes on DNA
  https://www.ncbi.nlm.nih.gov/books/NBK26872/#A1300

GeneReviews

- Rubinstein-Taybi Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1526

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28EP300%5BTI%5D%29+OR+%28E1A+binding+protein+p300%5BTI%5D%29%29+OR+%28%28E1A-binding+protein,+300kD%5BTI%5D%29+OR+%28p300%5BTI%5D%29+OR+%28p300+E1A-Associated+Coactivator%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D

OMIM

- COLORECTAL CANCER
  http://omim.org/entry/114500
- E1A-BINDING PROTEIN, 300-KD
  http://omim.org/entry/602700
- LEUKEMIA, ACUTE MYELOID
  http://omim.org/entry/601626
- PROSTATE CANCER
  http://omim.org/entry/176807

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/P300ID97.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=EP300%5Bgene%5D

- HGNC Gene Family: Lysine acetyltransferases
  http://www.genenames.org/cgi-bin/genefamilies/set/486
- HGNC Gene Family: Zinc fingers ZZ-type
  http://www.genenames.org/cgi-bin/genefamilies/set/91
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3373
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2033
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  http://www.uniprot.org/uniprot/Q09472

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https://ghr.nlm.nih.gov/gene/EP300

Reviewed: January 2007
Published: March 21, 2017

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